Indiana State Department of Health

Positive Cystic Fibrosis Newborn Screen – One Gene Change

What is newborn screening for cystic fibrosis?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to screen for a group of conditions. One of these conditions is **cystic fibrosis**, or CF.

My baby's screen was positive for one gene change. What does this mean?

Everybody inherits two copies of the CF gene (one from our mothers and one from our fathers). Sometimes these genes have **changes** (also called mutations) that prevent the gene from working correctly.

The CF newborn screen checks for 46 of the most common gene changes that cause CF. Your baby has a single copy of one of these gene changes.

M**ost children with one gene change will NOT have CF**. In order for a person to have CF, he or she must have two gene changes. People with one gene change are called "carriers."

What is a carrier?

People who have one gene change and one normal CF gene are called "**carriers**." Carriers do not have cystic fibrosis. However, carriers have a higher risk of having children with cystic fibrosis. If your child is a carrier of a CF gene change, it will be important for him or her to talk with a genetic counselor before they have children.

You may also meet with a genetic counselor to discuss the risk you and your partner have of having other children with CF. Other family members may also have a higher chance of having a child with CF. The genetic counselor will talk about these risks with you.

How will I know if my baby is a carrier?

Babies who have a positive CF newborn screen need to have a second test to find out whether they have cystic fibrosis.

This test is called a **sweat test**. The Indiana State Department of Health strongly recommends that your baby's sweat test be done at a laboratory accredited by the Cystic Fibrosis Foundation. Your baby's doctor will refer you to an accredited laboratory for the test.

What is a sweat test?

A **sweat test** checks to see if your baby has extra salt in their sweat. A small area of skin is prepped with a special chemical to make your baby sweat. This sweat will be collected on a piece of gauze and sent to the laboratory, where the amount of salt will be measured.

The sweat test takes about an hour. The results are usually available on the same day the test is done. No needles are used, and the test will not hurt your baby.

What should we do to prepare for the sweat test?

- Do not use any lotions or oils on your baby the day of the test. These may make it more difficult to collect sweat.
- Bring a blanket in case the testing center is cool.

I had a negative CF test during pregnancy. Does my baby still need a sweat test?

Yes. All babies that have a positive CF newborn screen need a sweat test.

Where can I get more information about cystic fibrosis?

- **Cystic Fibrosis Foundation**
  - www.cff.org
  - Toll-free (800) 344-4823
- **Indiana State Dept. of Health**
  - Toll-free (888) 815-0006
- **About Special Kids (ASK)**
  - www.aboutspecialkids.org
  - Toll-free (800) 964-4746